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(54) Title: FACL4 AND MUTATION THEREOF ON X-LINKED MENTAL RETARDATION SYNDROME

(57) Abstract: A nucleic acid comprising at least one fragment of the human FACL4 gene or FACL4 protein or functional portions thereof for diagnostic or therapeutic purposes applied to syndromes associated with mental retardation is described. Appropriate diagnostic kits are also described.

INTERNATIONAL SEARCH REPORT

International Application No

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A. CLASSIFICATION OF SUBJECT MATTER
 IPC 7 C12Q1/68 C12Q1/48

According to International Patent Classification (IPC) or to both national classification and IPC

B. FIELDS SEARCHED

Minimum documentation searched (classification system followed by classification symbols)
 IPC 7 C12Q

Documentation searched other than minimum documentation to the extent that such documents are included in the fields searched

Electronic data base consulted during the international search (name of data base and, where practical, search terms used)
 EPO-Internal, BIOSIS, WPI Data, PAJ, MEDLINE, EMBASE

C. DOCUMENTS CONSIDERED TO BE RELEVANT

Category	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
X	WO 02 16575 A (UNIV UTAH RES FOUND ;CAO YANG (US); PRESCOTT STEPHEN (US)) 28 February 2002 (2002-02-28) page 11, line 22-25; claims 25-28; example 1	1, 3-6, 9
X	CAO Y ET AL: "CLONING, EXPRESSION, AND CHROMOSOMAL LOCALIZATION OF HUMAN LONG-CHAIN FATTY ACID-COA LIGASE 4 (FACL4)" GENOMICS, ACADEMIC PRESS, SAN DIEGO, US, vol. 49, 1998, pages 327-330, XP002932974 ISSN: 0888-7543	3-8
Y	the whole document	11-13

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Patent family members are listed in annex.

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C.(Continuation) DOCUMENTS CONSIDERED TO BE RELEVANT

Category *	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
X	<p>PICCINI M ET AL: "FACL4, a New Gene Encoding Long-Chain Acyl-CoA Synthetase 4, Is Deleted in a Family with Alport Syndrome, Elliptocytosis, and Mental Retardation" GENOMICS, ACADEMIC PRESS, SAN DIEGO, US, vol. 47, no. 3, 1 February 1998 (1998-02-01), pages 350-358, XP002257224 ISSN: 0888-7543 cited in the application abstract Results figure 3</p>	3-8
X	<p>CAO YANG ET AL: "Intracellular unesterified arachidonic acid signals apoptosis" PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES, vol. 97, no. 21, 10 October 2000 (2000-10-10), pages 11280-11285, XP002257225 October 10, 2000 ISSN: 0027-8424 cited in the application abstract</p>	3-6
Y	<p>page 11280, column 2, paragraph 5 -page 11281, column 1, paragraph 1</p>	11-13
X	<p>MALHOTRA KIRAN T ET AL: "Identification and molecular characterization of acyl-CoA synthetase in human erythrocytes and erythroid precursors" BIOCHEMICAL JOURNAL, vol. 344, no. 1, 15 November 1999 (1999-11-15), pages 135-143, XP002257226 ISSN: 0264-6021 cited in the application page 137, column 1, paragraph 3</p>	13
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Y	<p>KANG MAN-JONG ET AL: "A novel arachidonate-preferring acyl-CoA synthetase is present in steroidogenic cells of the rat adrenal, ovary, and testis" PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES, vol. 94, no. 7, 1997, pages 2880-2884, XP002257227 1997 ISSN: 0027-8424 abstract</p>	11-13

C.(Continuation) DOCUMENTS CONSIDERED TO BE RELEVANT

Category °	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
X	<p>WILSON D B ET AL: "DISCOVERY OF AN ARACHIDONOYL COENZYME A SYNTHETASE IN HUMAN PLATELETS" JOURNAL OF BIOLOGICAL CHEMISTRY, vol. 257, no. 7, 1982, pages 3510-3515, XP002932976 ISSN: 0021-9258</p>	13
Y	<p>abstract page 3511, column 1, paragraphs 3,4</p>	11,12
A	<p>MINEKURA HIROYUKI ET AL: "Exon/intron organization and transcription units of the human Acyl-CoA synthetase 4 gene" BIOCHEMICAL AND BIOPHYSICAL RESEARCH COMMUNICATIONS, vol. 286, no. 1, 10 August 2001 (2001-08-10), pages 80-86, XP002257228 ISSN: 0006-291X the whole document</p>	1-13
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A	<p>SCHAFFER A J ET AL: "DNA VARIATION AND THE FUTURE OF HUMAN GENETICS" NATURE BIOTECHNOLOGY, NATURE PUBLISHING, US, vol. 16, January 1998 (1998-01), pages 33-39, XP000890128 ISSN: 1087-0156 abstract p.35, Box 1</p>	3-6
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Category *	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
P,X	LONGO I ET AL: "A third MRX family (MRX68) is the result of mutation in the long chain fatty acid-CoA ligase 4 (FACL4) gene: Proposal of a rapid enzymatic assay for screening mentally retarded patients." JOURNAL OF MEDICAL GENETICS, vol. 40, no. 1, January 2003 (2003-01), pages 11-17, XP002257230 ISSN: 0022-2593 the whole document	1-13

INTERNATIONAL SEARCH REPORT

patent family members

International Application No

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